

NNPDF-Funded Research Grant # 40

**TITLE: Lysosomal Storage Disease Research Consortium Contribution for
NNPDF to Join Consortium**

PROJECT INVESTIGATOR: Multiple - See "More Details"

PERIOD: 4/1/2004 - 3/31/2006

PROJECT DESCRIPTION

A consortium of Lysosomal Storage Disease support groups called the LSD Research Consortium is jointly funding research into issues of concern to all LSD's rather than having each disease group tackle these issues separately. The NNPDF contributed \$10,000 to this fund to become part of the LSDRC, allowing the NNPDF to have input on how funds will be used.

An example of a large issue of common concern include is translational research for improved delivery of therapeutic cells, proteins, genes, and small molecules across the blood-brain barrier. This is critical for those LSD's with neurological involvement (including NPA and NPC).

Encouraged by the formation of the LSDRC and other events showing LSD groups are working together, in July of 2004, the NIH issued a call for research applications on therapies for LSD's with neurological involvement called "PAS-04-120 - Central Nervous System THERAPY DEVELOPMENT FOR LYOSOMAL STORAGE DISORDERS".

See <http://grants.nih.gov/grants/guide/pa-files/PAS-04-120.html> for more details.

(Also see www.lsdresearch.org for more information on the Consortium.)

Final STATUS REPORT

Dated 1/25/2006

\$1,283,000 in spending on LSD research were awarded by the LSDRC and by NIH through PAS-04-120. The projects receiving grants were: Eain M. Comford, Ph.D, Professor of Neurobiology (David Geffen School of Medicine at UCLA) "Gene Delivery Across the Blood-Brain Barriocer in Lafora Knockout Mice" Philip E. Dawson, Ph.D, Assoc Professor, Cell Biology (Scripps Research Institute, LaJolla, California) "Potential of Chemical Chaperones and Thioester Reactive Small Molecules as Potential Therapeutic Approaches in the Treatment of Infantile Batten's Disease" Kostantin Dobrenis, Ph.D., Asst Professor, Neuroscience (Albert Einstein College of Medicine) "GM2 Gangliosidosis Therapy Using Neuronotropic Enzyme" Angela Gritti, Ph.D. (Institute for Stem Cell Research; Milan, Italy) "Neural Stem Cell Based Therapy for GM2 Gangliosidosis" Synthia Mellon, Ph.D., Professor, Ob/Gyn Reproductive Sciences (University of California at San Francisco) "Neurosteroid Therapy for Lysosomal Storage Disorders" Elizabeth Neufeld, Ph.D. (David Geffen School of Medicine at UCLA) "Aptamer-Directed Crossing of the

Blood Brain Barrier for Enzyme Therapy of LSD's" Thomas N. Seyfried, Ph.D., Professor of Biology (Boston College) "Evaluate MJ-DGJ as a substrate reduction therapy, neural stem cells (NSCs), as a cross-correctional therapy, and caloric restriction (CR) as an anti-inflammatory therapy for ganglioside storage diseases" Brian W. Soper, Ph.D., Research Staff Scientist (The Jackson Laboratory) "MPS VII CNS Gene Therapy Using Neuronal Stem Cells"

PUBLICATIONS:

No Publications on this Work To Date